KIF1B gene

kinesin family member 1B

Normal Function

The *KIF1B* gene provides instructions for making a protein called kinesin family member 1B, part of the kinesin family of proteins. These proteins are essential for the transport of materials within cells. Kinesin proteins function like freight trains that transport cargo, and their structure is suited for this cargo-carrying function. One part of the protein, called the motor domain, provides the power to move the protein and its cargo along a track-like system made from structures called microtubules. Another part of the kinesin protein, which varies among members of this protein family, binds to specific materials for transport.

Research suggests that the kinesin family member 1B protein specializes in carrying two types of cargo. In nerve cells (neurons), this protein transports small, sac-like structures called synaptic vesicles, which contain materials necessary for the transmission of nerve impulses. In other cell types, the kinesin family member 1B protein carries energy-producing structures called mitochondria.

In addition to its transport functions, the kinesin family member 1B protein appears to be involved in programmed cell death (apoptosis). Apoptosis is a common process throughout life that helps the body get rid of cells it does not need.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

One *KIF1B* gene mutation has been identified in a small number of individuals with a form of Charcot-Marie-Tooth disease known as type 2A. The mutation changes one of the protein building blocks (amino acids) in the motor domain of kinesin family member 1B. Specifically, the amino acid glutamine is replaced by the amino acid leucine at protein position 98 (written as Gln98Leu or Q98L). Although the effect of this mutation is not fully understood, it probably disrupts the transport of synaptic vesicles. A shortage of synaptic vesicles at nerve endings could impair the transmission of nerve impulses, causing the symptoms of type 2A Charcot-Marie-Tooth disease. However, some researchers question whether *KIF1B* gene mutations actually play a role in causing this disorder.

neuroblastoma

Deletion of a region of chromosome 1 containing the *KIF1B* gene, designated 1p36, has been identified in some people with neuroblastoma, a type of cancerous

tumor composed of immature nerve cells (neuroblasts). 1p36 deletions are somatic mutations, which means they occur during a person's lifetime and are present only in the cells that become cancerous. In addition, several inherited *KIF1B* gene mutations have been identified in families with a history of neuroblastoma. These mutations change single amino acids in the kinesin family member 1B protein. Studies suggest that deletion or mutation of the *KIF1B* gene may disrupt apoptosis, allowing cells to grow and divide too quickly or in an uncontrolled way. This kind of unregulated cell growth can lead to the formation of tumors.

nonsyndromic paraganglioma

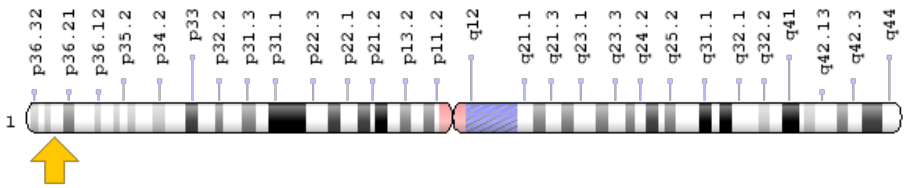
KIF1B gene mutations have been reported in individuals with a type of paraganglioma called pheochromocytoma. Paragangliomas are noncancerous (benign) tumors of the nervous system. Pheochromocytomas specifically affect the adrenal glands, which are small hormone-producing glands located on top of each kidney. These tumors often cause no symptoms, but in some cases they can produce an excess of hormones that cause dangerously high blood pressure. *KIF1B* gene mutations are associated with nonsyndromic pheochromocytoma, which means the tumors occur without additional features of an inherited syndrome.

The *KIF1B* gene mutations identified in nonsyndromic pheochromocytoma change single amino acids in the kinesin family member 1B protein. Studies suggest that the mutations may disrupt apoptosis, allowing cells to grow and divide too quickly or in an uncontrolled way and potentially leading to tumor formation.

Chromosomal Location

Cytogenetic Location: 1p36.22, which is the short (p) arm of chromosome 1 at position 36.22

Molecular Location: base pairs 10,210,706 to 10,381,603 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CMT2
- CMT2A
- HMSNII
- KIAA0591
- KIAA1488
- KIF1B_HUMAN
- KLP

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): There Are Two Types of Microtubule Motor Proteins: Kinesins and Dyneins
  https://www.ncbi.nlm.nih.gov/books/NBK26888/#A3047
- Molecular Cell Biology (fourth edition, 2000): Kinesin, Dynein, and Intracellular Transport
  https://www.ncbi.nlm.nih.gov/books/NBK21710/
- The Cell A Molecular Approach (second edition, 2000): Identification of Microtubule Motor Proteins
  https://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=cooper&part=A1833#A1834

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 2A
  https://www.ncbi.nlm.nih.gov/books/NBK1511

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28KIF1B%5BTIAB%5D%29+OR+%28kinesin+family+member+1B%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- KINESIN FAMILY MEMBER 1B
  http://omim.org/entry/605995

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_KIF1B.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KIF1B%5Bgene%5D
- HGNC Gene Family: Kinesins
  http://www.genenames.org/cgi-bin/genefamilies/set/622
- HGNC Gene Family: Pleckstrin homology domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/682
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16636
- Inherited Peripheral Neuropathies Mutation Database
  http://www.molgen.ua.ac.be/CMTMutations/Mutations/Contexts.cfm?ID=17
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/23095
- UniProt
  http://www.uniprot.org/uniprot/O60333

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https://ghr.nlm.nih.gov/gene/KIF1B

Reviewed: October 2011
Published: March 21, 2017

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